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IgG4 deficiency with gene deletion in Down syndrome

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Background & Aim: IgG4 deficiency is more frequent among persons with Down syndrome (DS), without identifying explanation. The role of IgG4 deficiency which is not fully established for many affected persons in the general population are asymptomatic. Nevertheless, in the context of DS it may be an important factor in repeated infections and even stroke. The aim of the present study was to investigate the molecular mechanism of IgG4 deficiency at the level of the heavy chain gene (IGHG4) gene.

Methodology: Quantitative real-time polymerase chain reaction (Q-PCR) was carried out to measure IGHG4 copies number with SYBR Green detection and comparison to a reference gene (36B4). A IGHG4/36B4 ratio was considered normal (2 copies of IGHG4) when between 0.8 and 1.2. We studied 44 DS persons: 21 males and 23 females from 7 years to 57 years, compos of 23 DS persons (11 males and 12 females) carrying severe IgG4 deficiency (<0.02 g /L), 5 having an IgG4 level not detectab and 21 DS subjects (10 males and 11 females) with no IgG4 deficiency (level >0.1 g /L). The patient group was compared with 38 healthy donors (controls) without DS.

Results:IGHG4 heterozygous deletion was found in 16 (69.6%) DS patients with IgG4 deficiency versus in 2 (9.5%) DS subjects without IgG4 deficiency (p=0.0001 with Yates correction) in the control group, no deletion was seen.

Conclusions:IGHG4 haploinsufficiency is highly correlated to IgG4 deficiency in our population with DS, but other factors exist that needs to be identified.

Biography

Jeraiby M has completed his residency in Medical Biochemistry at Saint-Étienne University Hospital Center, France and has been working as a Professor of Medical Biochemistry at Jazan University, Saudi Arabia. He has published many papers in reputed journals and has been serving as a Reviewer in clinical case report journal.

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